The Haplotype Map Project (HapMap)
and Other Research on Genetic Variations

Please take as much time as you want to read this form, ask questions, and talk about this project with family or friends.

What is this project about?

The National Institutes of Health (NIH), the main U.S. government agency that funds medical research, is collecting blood samples from people whose ancestors came from different parts of the world. About 100-200 people from each of about 10 ethnic or geographic groups will give samples for this project. The University of Utah is helping the NIH with this project. We will not collect new samples, but we are asking for your consent to have your existing sample used in this project and in future related projects.

Why is the NIH doing this project?

Genes are the basic "instruction book" for people. Genes are made out of DNA. The DNA of a person is about 99.9% the same as the DNA of any other person. But no two people have exactly the same DNA except identical twins. Differences in DNA are called genetic variations. They explain some of the differences among people, like eye colors and blood groups. They also partly explain why some people get diseases like cancer, diabetes, asthma, and depression, while others do not. Diseases such as these are also affected by diet, exercise, smoking, pollution, and other factors, which makes it hard to figure out which genes affect the diseases.

Most genetic variations are found in people everywhere. But there are differences among groups in how common some genetic variations are. For example, all the blood types can be found in all groups of people in every part of the world, but there are differences from place to place in how often some blood types appear. By including people from many ethnic and geographic groups, researchers will find most of the genetic variations. This information will make it easier for researchers to find the genes that affect diseases, in other studies using other samples.

How will the samples be used?

Over the next three years, researchers will study the samples to find places in the DNA where people vary. For each sample, researchers will make a list of the genetic variations they find. Researchers will also look for the patterns of genetic variations in people's DNA, which are called "haplotypes."

Researchers will put all this information in a scientific database on the Internet. For each sample, this will include information on hundreds of thousands (eventually millions) of genetic variations, as well as the ethnic or geographic group, the sex, and the position in the family of the person who gave the sample. The database will not include any medical information about anyone whose sample is used. It also will not include any information that could identify who the individual people or families are.

Researchers will use the genetic variation information in the database to create a genetic map that summarizes the patterns of genetic variation, called a haplotype map or "HapMap." The HapMap will be put on the Internet. The HapMap will not include medical information, but researchers will use it as a tool in future studies to find genes related to many diseases. The HapMap will show researchers where

the common haplotypes (patterns of genetic variation) are. Then, for a disease, such as diabetes, researchers will study the haplotypes in a group of people who have the disease, and in another group of people who do not. Areas in the DNA where the two groups differ in their haplotypes will be clues that those areas might contain genes that affect the disease. Researchers can then look for those genes and study how they work. This will help them figure out better ways to prevent, diagnose, and treat the disease. They can also learn how to make drugs that work better in more people. Some researchers will also use the HapMap to look for genes that affect traits such as baldness, behaviors like addiction, and long life.

Researchers will compare the genetic variation and haplotype information for people in the same group and in different groups. In the future, researchers will also use the existing cell line samples to look for differences in the amount and form of the products that genes make, called RNA and proteins, and will put all this information for each sample in the database. The samples, the database, and the HapMap will also be used to study other questions, such as the biology of DNA, how new variations arise, the genetic history of human groups, and how people from different parts of the world are related to each other.

Because the database will be public, people who do identity testing, such as for paternity testing or law enforcement, may also use the samples, the database, and the HapMap, to do general research. *However, it will be very hard for anyone to learn anything about you personally from any of this research because none of the samples, the database, or the HapMap will include your name or any other information that could identify you or your family.*

What will happen if I agree to let my sample be used for this project?

Your sample will continue to be stored at the University of Utah, at the Centre d'Etude du Polymorphisme Humain (CEPH) in Paris, France, and at the Human Genetic Cell Repository at the Coriell Institute for Medical Research in New Jersey (the "Repository"), which the NIH oversees. Your sample is being stored as a cell line, which makes an unlimited amount of your DNA and will last a long time.

The Repository will send the cell lines to researchers around the world to create the HapMap and to use in many future genetic studies as described in this form. The researchers will have to follow all U.S. and international laws and guidelines that apply to research. All studies using the cell lines from the Repository will have to be approved by the Institutional Review Board (IRB) of the Repository. An IRB is a committee similar to the one that approved this project to make sure that your rights were protected.

What will happen if I don't agree to let my sample be used?

You will not lose any benefits if you choose not to let your sample be used. If you don't agree to let your sample be used, it will not be used for the HapMap. However, it will continue to be used for other IRB-approved research studies, just as it has been in the past, unless you specifically tell us that you don't want it used for such studies anymore.

Will there be any costs or payments?

It will not cost you anything to have your sample used for this project.

The Repository does not let anyone sell material from samples or cell lines. However, information from genetics research sometimes helps companies make products to diagnose or treat diseases. If information from your family's cell lines leads to making a product, it would probably contribute only in a very small

way. Also, because the cell lines will not have names on them, neither the researchers nor anyone at the Repository would know if your samples were even used. So you will not get any additional payment for having your sample used in this project.

How will you protect my privacy?

We will protect your privacy carefully, just as we have always done in the past. The only people who will know your name or any other personal identifying information will be the clinic coordinator, the physician, and the principal investigator for the Utah Genetic Reference Project at the University of Utah. We will not give this information to anybody else. While the University of Utah will keep your new, signed consent form, nobody else will see it. The sample stored at the Repository and used for the HapMap will not have your name on it. Although it will have a code number, nobody except us will know the name of the person the code number is linked to. So nobody at the Repository or who studies your sample will know that it came from you.

While we are asking everyone who originally gave us a sample to consent to letting that sample be used in this project, not everybody's sample will be used for the HapMap. So nobody, not even you or us, will know if your sample was used for the HapMap.

What are the benefits of having my sample used for this project?

You will not directly benefit from having your sample used for this project because of the long time this research will take to produce useful results. But researchers will study these samples for many years to learn about health and disease. This research will eventually benefit the health of people around the world.

What are the risks of having my sample used for this project?

If your family's samples are used, lots of genetic information from your samples will be put in the database, and lots of people will be able to look at it for any purpose. However, there are only a couple of ways anybody could trace the information back to you. One is if they thought your information might be in the database, got another sample from you, did many tests on that sample, and then compared the genetic information from those tests with the information in the database. The other is if somebody compared the information in the database with genetic information known to be from you that was in another database and figured out who you were. The risk of either of these things happening is very small, but it may grow in the future.

We cannot always predict the results of research, so new risks to you may come up in the future that we can't predict now.

Are there any risks to the community?

Information on the ethnic or geographic groups the samples came from will be included with the samples, in the database, and in the HapMap. In future studies, researchers may find that certain genetic variations appear more often in people from your community than in people from other groups, and that these variations are more common in people with a certain disease. This may make some people look down on your community unfairly.

Some people may use the information from the HapMap or from future studies using the HapMap to exaggerate differences between groups for prejudiced or other bad reasons. Others may use the

information to downplay differences between groups, to say that all people's genes are about the same, so we don't need to respect the special concerns of different groups. Biology does not provide a reason for prejudice, but discrimination does exist.

We will work to make sure that the ethnic or geographic identity of your community is described as carefully as possible--in the sample collection, in the database, in the HapMap, and in any articles researchers write about the HapMap. Your sample will continue to be part of the Utah Genetic Reference Project. The term "CEPH" (Centre d'Etude du Polymorphisme Humain) may sometimes be used to name this collection.

Can I change my mind after I agree to let my sample be used?

Deciding whether to let your sample be used for the HapMap is completely up to you. You will not lose any benefits if you choose not to let your sample be used. However, once your sample has been studied and your genetic information has been put in the database, you will not be able to take that information back.

How will I find out what happens with this project?

We will not be able to give you any individual results from this research. However, we will update your community from time to time on how researchers are using the HapMap and your community's samples and what they are learning about health and disease.

Who can I talk to if I have questions or problems?

If you have questions about this re-consent process, contact:
Missy Dixon, Study Coordinator (801) 585-6807; email: missy.dixon@genetics.utah.edu
Dr. Mark Leppert, Principal Investigator: (801) 581-8131

If you have questions regarding your rights as a research subject, or if problems arise which you do not feel you can discuss with the Investigator, please contact the Institutional Review Board Office at (801) 581-3655. (University of Utah)

Medical Treatment or Compensation for Injury:
In the event you sustain injury resulting from your participation in the research project, the University of Utah can provide to you, without charge, emergency and temporary medical treatment not otherwise covered by your own insurance. If you believe that you have sustained an injury as a result of your participation in this research program, please contact the Institutional Review Board, phone number (801) 581-3655. By signing this document you are not giving up your right to pursue legal action against any and all parties involved with this research, in accordance with the Utah Governmental Immunity Act, Section 63-30-1:63-30-34 Utah Code Ann. 1953 (as amended).

Consent and Signature

Please read the paragraph below, think about your choice, and sign if you agree:

I agree to have my cell line used for the HapMap Project and in other approved studies of the type described in the form. I have read or listened to the information, I have asked any questions I had, and all my questions were answered. I know that re-consenting to the use of my sample for this project is my choice. I understand that after I agree to let my cell line be used for the HapMap, I cannot change my mind.

Please initial box

1. I confirm that I have read and understand this consent document and have had the opportunity to ask questions. ❑

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected. (Please refer to page 4, paragraph 2). ❑

3. I understand that sections of any of my medical notes may be looked at by responsible individuals from the University of Utah or from regulatory authorities where it is relevant to my taking part in research. I give permission for these individuals to have access to my records. ❑

4. I agree to take part in the above study and that I will be given a signed copy of the consent form to keep. ❑

Signature Block: If the subject is competent to give consent, the form must be signed and dated. Also, a witness must sign and date the form after having watched the consent process. The investigator must keep a signed copy of the IRB approved consent form with the IRB approved date and expiration date. If the subject is not competent to give consent, the form must be signed and dated by the subject's legal representative/next of kin.

_____ _____

Signature of Participant Date

_____ _____

Signature of Witness Date

_____ _____

Signature of Researcher or staff Date

Copy given to participant: _____Yes